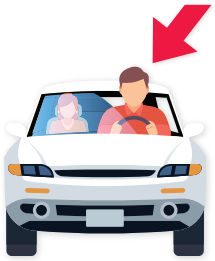
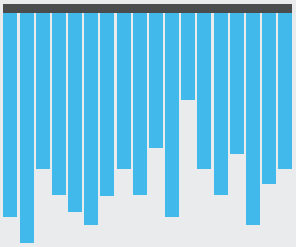
UNRAVELING THE COMPLEXITIES OF CANCER GENOMICS

Global efforts from an international team of researchers have led to one of the most comprehensive studies of the whole genome of more than 2,600 tumors from 38 different types of cancer. Among the most important findings, published recently, were the following:

Most tumors contain at least one identifiable mutation in their genomes that appears to drive tumor growth and on an average each cancer genome was found to contain between four and five of such "driver" mutations. Interestingly, some of these mutations were detected in parts of the DNA referred to as "noncoding regions" which have traditionally not been a focus of cancer research. These discoveries are a major stride toward cataloging important cancer-causing genetic changes, which is critical for the advancement of precision medicine.



Unique patterns of mutations referred to as "mutational signatures" are often associated with processes that may lead to cancer development, such as defective DNA-repair mechanisms or exposure to cancer risk factors such as environmental carcinogens, toxicants in tobacco smoke, or ultraviolet radiation. Collectively, the researchers identified 97 signatures from a variety of tumors. Notably, the causes of many such signatures were unknown, suggesting that more work needs to be done to identify currently unrecognized cancer risk factors.



By analyzing the vast array of genetic changes, the researchers were able to determine the chronology of cancer-causing mutations. They found that many mutations can occur years, if not decades, prior to a cancer diagnosis. These findings have potentially important implications for early detection and interception of these cancers.

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